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158340**[Links](#)**MUCIN 1, TRANSMEMBRANE; MUC1Alternative titles; symbols*


**MUCIN 1, URINARY
PEANUT-REACTIVE URINARY MUCIN; PUM
MUCIN, TUMOR-ASSOCIATED EPITHELIAL
POLYMORPHIC EPITHELIAL MUCIN; PEM**


Gene map locus 1q21**TEXT**

Karlsson et al. (1983) demonstrated a genetically determined polymorphism of a human urinary mucin by the separation technique of SDS polyacrylamide gel electrophoresis followed by detection with radioiodinated lectins. Peanut agglutinin was the most effective lectin; hence, the proposed designation peanut-reactive urinary mucin (PUM). Karlsson et al. (1983) identified 4 common alleles with codominant inheritance. The same polymorphic protein is expressed in other normal and malignant tissues of epithelial origin including the mammary gland. Variation in white cell DNA detected with a cDNA probe for mammary mucin exactly matches the variation of the protein as demonstrated after electrophoresis using a series of monoclonal antibodies; studies in 2 large families demonstrated the precise correspondence. It appears that a series of tandem repeats constitutes much of the coding region of the PUM gene and that the allelic variation is due to variation in the number of repeats, as occurs in the hypervariable minisatellite regions of DNA. Swallow et al. (1987) mapped the PUM locus to 1q21-q24 by somatic cell hybrid studies and in situ hybridization. Because of its high order of heterozygosity, the PUM locus should be exceedingly useful for mapping 1q. Indeed, Swallow et al. (1987) presented evidence obtained using a cDNA that the PUM locus is a hypervariable 'minisatellite' region similar to those described by several groups, but novel in that it is transcribed and translated, and that the same polymorphism is demonstrable in the expressed gene product. Swallow et al. (1988) found close linkage of Duffy blood group (110700) and PUM (maximum lod score = 4 at theta = 0). Middleton-Price et al. (1988) found linkage of alpha-spectrin (182860) and PUM (maximum lod score = 5.98 at theta = 0.05); both loci may lie within 1q21. Anderson et al. (1989) presented results of linkage studies of PUM and chromosome 1 markers in the CEPH families. Gendler et al. (1990) reported the full sequence of epithelial mucin as deduced from cDNA sequences. Length variations in the tandem repeat result in MUC1 being an expressed variable number tandem repeat (VNTR) locus. Tandem repeats appear to be a general characteristic of mucin core proteins. Gendler et al. (1990) studied the polymorphic epithelial mucin present on the surface of human mammary cells. It is developmentally regulated and aberrantly expressed in breast cancer. Lan et al. (1990) used a monospecific polyclonal antiserum against deglycosylated human pancreatic tumor mucin to select clones from a cDNA library developed from a human pancreatic tumor cell line. The close similarity of the cDNA sequence and the deduced amino acid sequence of pancreatic mucin to those of breast tumor mucin, as reported by

Gendler et al. (1990) and others, led them to suggest that the core protein, the apomucin, is produced by the same gene. The native forms of these molecules are distinct in size and degree of glycosylation, however, suggesting that factors other than the primary structure of the apomucin determine these characteristics. The many complex O-linked oligosaccharides (which make up to 80% of the total molecular weight) are presumably added by glycosyltransferases. Lan et al. (1990) found that the structure of pancreatic tumor mucin is quite different from that of intestinal mucin (158370). 

By analysis of interspecific backcross mice, Kingsmore et al. (1995) mapped the homologous gene to mouse chromosome 3.

A polymorphism due to a G/A substitution in exon 2, responsible for a genetically determined variation in splicing of the MUC1 transcript, was reported by Ligtenberg et al. (1990); Ligtenberg et al. (1991). Pratt et al. (1996) reported a CA repeat polymorphism within intron 6 of the gene. The various results supported the notion that the VNTR polymorphism in the coding sequence of MUC1 was not caused by unequal reciprocal recombination at meiosis. 

Silva et al. (2001) evaluated the MUC1 VNTR polymorphism in a series of 174 patients with chronic gastritis in a population from northern Portugal with a high incidence of gastric carcinoma (137215). The data were compared with those from blood donors and patients with gastric cancer from the same population. Significant differences were observed between patients with chronic gastritis and blood donors and also between patients with chronic gastritis and patients with gastric cancer. Homozygotes for small MUC1 VNTR alleles were significantly associated with gastric carcinoma as well as with chronic atrophic gastritis and incomplete intestinal metaplasia, the 2 well established precursor lesions of gastric carcinoma, suggesting that MUC1 genotypes may define different susceptibility backgrounds in the gastric carcinogenesis pathway. 

SEE ALSO

Ligtenberg et al. (1991); Swallow et al. (1987); Swallow et al. (1987)

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